New drug moves closer to becoming first treatment for Fragile X Syndrome

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A new drug discovered through a research collaboration between the University at Buffalo and Tetra Therapeutics took a major step toward becoming a first-in-class treatment for Fragile X Syndrome, a leading genetic cause of autism.

The drug, BPN14770, achieved positive topline results in a phase 2 clinical study. The innovative treatment improved cognitive function in adult male patients with Fragile X Syndrome.

Fragile X Syndrome—a genetic disorder for which there is no cure—is the most commonly known cause of inherited intellectual disability, according to the Centers for Disease Control and Prevention.

"We are very excited about the results of this study," said Mark Gurney, Ph.D., founder and chief executive officer of Tetra Therapeutics. "In addition to being safe and well tolerated, treatment with BPN14770 led to significant cognitive improvement, specifically in the language domains, and we also saw a clinically meaningful benefit in overall daily functioning. These findings validate our approach to treating this disease through a mechanism that addresses a core deficit in the disorder."

The research was conducted at Rush University Medical Center by principal investigator and pediatric neurologist Elizabeth Berry-Kravis, MD, Ph.D. Funding was provided by the FRAXA Research Foundation, a nonprofit dedicated to financing Fragile X Syndrome research.

Preclinical investigation of BPN14770 was completed through a collaboration between UB School of Pharmacy and Pharmaceutical Sciences faculty members James M. O'Donnell, Ph.D., dean and professor, and Ying Xu, MD, Ph.D., research associate professor, and biotechnology company Tetra Therapeutics.

The drug inhibits the activity of phosphodiesterase-4D, an enzyme that plays a key role in memory formation, learning, neuroinflammation and traumatic brain injury. Previous studies found that BPN14770 has the potential to promote the maturation of connections among neurons, which are impaired in patients with Fragile X Syndrome.

"The collaboration with Tetra Therapeutics has been interesting and productive, combining our lab's expertise in preclinical pharmacology and theirs in drug discovery and development," said O'Donnell. "Seeing years of research lead to a successful trial for treatment of this serious genetic disorder is quite rewarding."

BPN14770's potential to improve cognitive and memory function could also translate to treatments for Alzheimer's disease, developmental disabilities, traumatic brain injury and schizophrenia.

Provided by University at Buffalo